SAMPLE LIST OF QUESTIONS TO EXAM ON MEDICAL GENETICS

List of exam questions discussed and approved at faculty meeting from 03.11.2022, Protocol No. 2

GENERAL ISSUES OF MEDICAL GENETICS

1. "Four P" in modern medicine: predictive (predictive), preventive (pre-emptive), personalized, participatory (providing for the active role of the patient).
2. Accumulation diseases (mitochondrial, lysosomal, peroxisomal).
3. Diseases with hereditary predisposition.
4. Diseases of expansion and the concept of the genetic load of the population.
5. Gallinowski direction in the development of genetics, eugenics and neoeugenics, their criticism.
6. Genetic and protein markers in the diagnosis of genetic and hereditary diseases.
7. Watch genetic migration and settlement of humanity. Role in the formation and population accumulation of orphan diseases. The role of the bottleneck effect in the accumulation of recessive pathological mutations in the population. The founder effect (founder effect).
8. Genetic mosacism. Examples of chromosomal and gene pathologies occurring against the background of genetic mosaicism.
9. Gene pool as a strategic resource. Genetic prediction. The concept of genofond.
10. Classification of hereditary diseases (base OMIM, Paris classification).
11. Classification of mutations.
12. Clinical and genetic study, indications for it.
13. Medical and genetic counseling, its goals, objectives, stages (phenotyping, setting clinical and genealogical pedigree, medical and genetic conclusion), level of organization and legal framework. Indications for medical and genetic counseling.
14. Methods of genetic analysis (cytogenetic, molecular cytogenetic, molecular genetic, clinical and genealogical).
15. PCR methods in medical genetics.
16. Microsimulations syndrome – General characteristics.
17. Mitochondrial inheritance. Differences of cytoplasmic inheritance from nuclear.
18. Mitochondrial segregation and its effects.
19. Monogenic hereditary diseases-General characteristics.
20. Multifactorial hereditary diseases-General characteristics.
21. Heredity and variability. Types of variability, the role of environmental factors in the formation of phenotypic variability.
22. Hereditary cancer syndromes-General characteristics.
23. Features manifestations of hereditary diseases. Terms of manifestation of chromosomal, monogenic and multifactorial diseases.
24. Risk assessment of hereditary diseases.
25. Indications for referral to the diagnosis of metabolic disorders in children 1st and 2nd year of life.
26. Polygenic and multifactorial principles of phenotype formation. The role of habitat factors in the formation of healthy and painful phenotype.
27. The concept of orphan diseases. Diagnosis, treatment, ethical aspects of the diagnosis of orphan diseases.
28. Postnatal diagnosis of hereditary and congenital diseases.
29. Predictive medicine. Proteomic analysis.
30. Pre-conception prevention of gene diseases.
31. Prenatal diagnosis of hereditary and congenital diseases.
32. Prevention of hereditary diseases.
33. Semiotics of genetic diseases.
34. Screening programs as prevention of congenital and hereditary diseases.
35. Types of inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked, mitochondrial. Brief examples of pedigrees.

36. Mental retardation in hereditary diseases (chromosomal syndromes, metabolic syndromes-examples of disorders of genes and proteins encoded by them). Proteins of the CE matrine, synuclein, nerve growth factor and their genes.
37. Pharmacogenetics, stages of xenobiotic biotransformation and examples of their disorders. The system of nonspecific oxidases and its relation to oncogenesis.
38. Small anomalies of development and their characteristics. Teratogenesis.
39. Etiology and pathogenesis of mitochondrial diseases.
40. Etiology and pathogenesis of lysosomal diseases.
41. Etiology and pathogenesis of monogenic metabolic diseases.
42. Etiology and pathogenesis of peroxisomal diseases.
43. Etiology and pathogenesis of chromosomal diseases.
44. Ethical issues of medical genetic counseling.

PRIVATE ISSUES OF MEDICAL GENETICS

CLINIC AND DIAGNOSTICS:

Chromosomal syndromes:

45. Down's disease, typical and atypical forms.
46. Klinefelter syndrome: clinical variants.
47. Shereshevsky-Turner syndrome: clinical variants.
48. Patau syndrome.
49. Trisomy x syndrome.
50. Edwards syndrome.
51. Cat's cry syndrome.

Monogenic diseases:

52. Lethal osteopetrosis,
53. Congenital isolated hypotrichosis.
54. Hyperhomocysteinemia. Clinical significance in the practice of medical geneticist (neurologist), cardiologist and obstetrician-gynecologist.

Monogenic diseases detected during newborn screening:

55. Adrenogenital syndrome.
56. Gaucher disease.
57. Tay-Sachs disease.
58. Congenital hypothyroidism.
59. Galactosemia.
60. Duchenne, Becker myodystrophy.
61. Cystic fibrosis.
62. Phenylketonuria.
63. Celiac disease.

Polygenic diseases:

64. Schizophrenia.
65. Epilepsy.
66. Hereditary forms of dementia.

(Type II diabetes, hypertension, asthma, osteoporosis, meta-Bolic syndrome are considered in the course of internal medicine).

Diseases of blood and cellular immunity system:

67. Congenital erythrocytosis.
68. Hereditary hemolytic anemia.

69. Some forms of congenital immunodeficiency.
70. Erythropoietic uroporphyria (Gunter's disease) and protoporphyria.

Mitochondrial diseases:
71. Mitochondrial diseases associated with the pathology of the Krebs cycle enzymes.
72. Mitochondrial diseases associated with the pathology of respiratory chain enzymes.
73. Mitochondrial diseases caused by changes in the number of copies of the mitochondrial chromosome.
74. The mucopolysaccharidoses (Hurler, Siegfried, Sanfilippo, etc.): clinic, diagnostics.

Peroxisome diseases:
75. Adrenoleukodystrophy.
76. Zellweger syndrome.
77. Refsum disease.
78. Gilbert syndrome.

Congenital connective tissue pathology:
79. Marfan syndrome.
80. Ehlers-Danlo syndrome.

Neurological diseases and syndromes:
81. Wilson-Konovalov disease.
82. Niemann-Pick disease.
83. Parkinson's disease.
84. Fabry's disease.
85. Thomsen congenital myotonia.
86. Crabbe-Beneke leukodystrophy.
87. The leukoencephalopathy of Pelizeus-Merzbacher.
88. Myotonic dystrophy of Churchman-batten-Steinert.
89. Charcot-Marie-Tuta neural amyotrophy.
90. Gilles de La Tourette syndrome.
91. Spinal amyotrophy of Verdnik-Hoffman, Kugelberg-Welander.
92. Charcot-Marie-Tuta neural amyotrophy.
93. Spinocerebellar degeneration. Friedreich's disease.
94. Olivo-Ponto-cerebellar atrophy.
95. Huntington's chorea.

Phakomatoses:
96. Ataxia-telangiectasia.
97. Neurofibromatosis.
98. Tuberous sclerosis, a disease of Bourneville of Pringle.
99. Hippel-Lindau cerebroretinal angiomatosis.
100. Sturge-Weber encephalotrigeminal angiomatosis.